PDE6H gene

phosphodiesterase 6H

Normal Function

The *PDE6H* gene provides instructions for making one part (called the inhibitory gamma subunit) of an enzyme called cone-specific phosphodiesterase. This enzyme is found exclusively in light-detecting (photoreceptor) cells called cones, which are located in a specialized tissue at the back of the eye known as the retina. Cones provide vision in bright light (daylight vision), including color vision. Other photoreceptor cells, called rods, provide vision in low light (night vision).

When light enters the eye, it stimulates specialized pigments in photoreceptor cells. This stimulation triggers a series of chemical reactions that produce an electrical signal, which is interpreted by the brain as vision. This process is called phototransduction. Cone-specific phosphodiesterase carries out one of the reactions in this process. Specifically, the enzyme converts a molecule called cGMP to another molecule, 5'-GMP, in cones. This conversion causes certain channels on the cell membrane to close. The closing of these channels triggers the transmission of visual signals to the brain.

Health Conditions Related to Genetic Changes

achromatopsia

At least one mutation in the *PDE6H* gene has been found to cause the vision disorder achromatopsia. It is a very rare cause of a form of the disorder called incomplete achromatopsia. This condition is characterized by limited color vision and other vision problems that are present from early infancy.

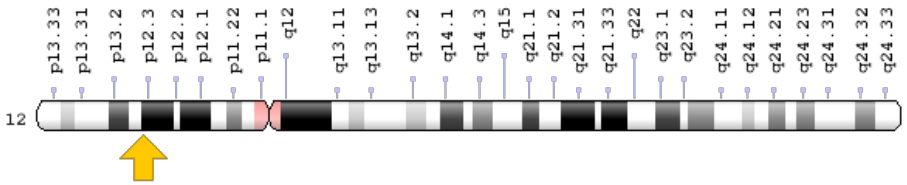
The identified mutation replaces one protein building block (amino acid) near the beginning of the inhibitory gamma subunit with a signal to stop protein production. This mutation is written as Ser12Ter or S12X. This change prevents the production of any functional inhibitory gamma subunit, which interferes with the normal function of cone-specific phosphodiesterase. Impairment of this enzyme disrupts the process of phototransduction in cones; rods are typically unaffected.

Because cones are needed for color vision, affected individuals have difficulty distinguishing certain colors. Their other vision problems are also related to the malfunctioning cones.

Chromosomal Location

Cytogenetic Location: 12p12.3, which is the short (p) arm of chromosome 12 at position 12.3

Molecular Location: base pairs 14,973,022 to 14,982,697 on chromosome 12 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ACHM6
- GMP-PDE gamma
- phosphodiesterase 6H, cGMP-specific, cone, gamma
- RCD3
- retinal cone rhodopsin-sensitive cGMP 3',5'-cyclic phosphodiesterase subunit gamma

Additional Information & Resources

Educational Resources

- Basic Neurochemistry (sixth edition, 1999): Cyclic Nucleotide Phosphodiesterases https://www.ncbi.nlm.nih.gov/books/NBK27996/
- Neuroscience (second edition, 2001): Phototransduction https://www.ncbi.nlm.nih.gov/books/NBK10806/
- Webvision: The Organization of the Retina and Visual System (2010): Phototransduction in Rods and Cones https://www.ncbi.nlm.nih.gov/books/NBK52768/

GeneReviews

- Achromatopsia https://www.ncbi.nlm.nih.gov/books/NBK1418

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28PDE6H%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D

OMIM

- PHOSPHODIESTERASE 6H, cGMP-SPECIFIC, CONE, GAMMA
  http://omim.org/entry/601190

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=PDE6H%5Bgene%5D
- HGNC Gene Family: Phosphodiesterases
  http://www.genenames.org/cgi-bin/genefamilies/set/681
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=8790
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/5149
- UniProt
  http://www.uniprot.org/uniprot/Q13956

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